CR-GE-45

XO/XY MOSAICISM IN AN 18-YEAR-OLD GIRL WITH PRIMARY AMENORRHEA

https://doi.org/10.15605/jafes.034.02.S137

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INTRODUCTION
45XO/46XY mosaicism is rare and may present with a wide spectrum of phenotypes which may go unrecognized. Y chromosome material has been shown to increase the risk for gonadal malignancy with gonadoblastoma being the most common germ cell tumour. Prophylactic gonadectomy has been recommended in all female individuals with Y chromosome material identified on standard karyotyping although it is debatable whether this may represent over-treatment.

CASE
We report an 18-year-old girl who was referred to endocrine clinic for primary amenorrhea. She is 145 cm tall with a mid-parental height of 152 cm. Breast development was Tanner 3 and pubic hair was Tanner 2. There were no obvious physical features of Turner syndrome, clitoromegaly or palpable gonads.

CONCLUSION
There are no guidelines on identifying the malignancy risk or timing of gonadectomy in patients with XY gonadal dysgenesis. This case illustrates the challenges in surveillance for these patients. A careful review of the physical features, hormonal evaluation, karyotype and malignancy risk should be undertaken and the findings discussed with the patient and family.

KEY WORDS
mixed gonadal dysgenesis, Turner syndrome, y chromosome, sex chromosome, aberrations, 45x/46y mosaicism

CR-GE-46

NONVIRILIZED: A CASE REPORT OF KALLMANN SYNDROME

https://doi.org/10.15605/jafes.034.02.S138

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INTRODUCTION
Kallmann Syndrome is a rare genetic disorder characterized by hypogonadotropic hypogonadism and hyposmia or anosmia due to mutations in one or more genes associated with olfactory bulb morphogenesis and the migration of GnRH neurons from their origin in the region of the olfactory placode.

CASE
A 25-year-old male initially consulted due to infertility. He had eunuchoid body proportion, high-pitched voice, absence of facial, axillary hair and pubic hair and small-sized penis. Laboratory work-up revealed an inappropriately normal serum luteinizing hormone and decreased serum testosterone level. Cranial MRI with contrast revealed empty sella. He was given supplementary Testosterone injection. At this time, he noticed development of sparse axillary hair, pubic hair, and decreased tone of voice. Examination of the genitalia revealing penis and testes at Tanner Stage III. He is currently receiving his testosterone injections which are 8 weeks apart aimed at maintaining his testosterone level at the mid normal range.

CONCLUSION
A thorough history and physical examination is needed and supplemented with appropriate diagnostic examinations for proper management of our patient. Although a rare disorder, we must include this differential diagnosis in approaching the case.

KEY WORDS
kallman, reproductive, infertility